



CYP11B2 gene

cytochrome P450 family 11 subfamily B member 2

Normal Function

The *CYP11B2* gene provides instructions for making an enzyme called aldosterone synthase (previously known as corticosterone methyloxidase). This enzyme is found in the adrenal glands, which are located on top of the kidneys. Aldosterone synthase is a member of the cytochrome P450 family of enzymes. These enzymes are involved in the formation and breakdown of various molecules within cells.

Aldosterone synthase helps produce a hormone called aldosterone. Aldosterone helps control blood pressure by maintaining proper salt and fluid levels in the body. The aldosterone synthase enzyme is involved in a series of three chemical reactions that produce aldosterone from other (precursor) molecules: the conversion of 11-deoxycorticosterone to corticosterone, the conversion of corticosterone to 18-hydroxycorticosterone, and the conversion of 18-hydroxycorticosterone to aldosterone.

Health Conditions Related to Genetic Changes

corticosterone methyloxidase deficiency

At least 30 *CYP11B2* gene mutations that cause corticosterone methyloxidase deficiency (also known as aldosterone synthase deficiency) have been identified. These mutations lead to insufficient production of aldosterone, which impairs the kidneys' ability to reabsorb salt (sodium chloride or NaCl) into the blood and release potassium in the urine. As a result, excessive amounts of salt in the form of charged atoms (ions) of sodium (Na^+) and chlorine (Cl^-) leave the body in the urine, while not enough potassium is released. The resulting imbalance of ions in the body underlies the signs and symptoms of this disorder, which include nausea, vomiting, dehydration, low blood pressure, extreme tiredness (fatigue), and muscle weakness.

familial hyperaldosteronism

A genetic change affecting the *CYP11B2* gene causes familial hyperaldosteronism type I, a disorder that leads to high blood pressure (hypertension). This change joins (fuses) the section of the *CYP11B2* gene that contains the instructions for making aldosterone synthase to a section of a nearby gene called *CYP11B1*. The added part of the *CYP11B1* gene contains a section called a promoter region, which normally helps start the production of an enzyme called 11-beta-hydroxylase from the *CYP11B1* gene.

By binding to the *CYP11B1* gene's promoter region, a hormone called adrenocorticotrophic hormone (ACTH) normally triggers production of the 11-beta-hydroxylase enzyme. In the fusion gene, ACTH binding abnormally triggers production of aldosterone synthase. High levels of aldosterone synthase result in excessive aldosterone production, which leads to the hypertension associated with familial hyperaldosteronism type I.

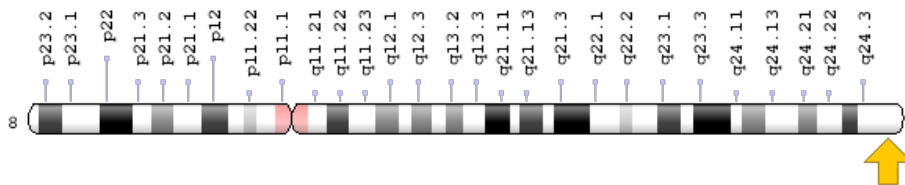
other disorders

Normal variations (polymorphisms) of the *CYP11B2* gene have been associated with increased risk of cardiovascular problems including hypertension, stroke caused by a lack of blood flow to the brain (ischemic stroke), and a heart rhythm abnormality called atrial fibrillation in people with heart failure. Researchers suggest that differences in blood pressure control resulting from the *CYP11B2* gene variations may lead to increased risk of these cardiovascular problems.

Chromosomal Location

Cytogenetic Location: 8q24.3, which is the long (q) arm of chromosome 8 at position 24.3

Molecular Location: base pairs 142,910,559 to 142,917,843 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ALDOS
- aldosterone synthase
- aldosterone-synthesizing enzyme
- C11B2_HUMAN
- CPN2
- CYP11B
- CYP11BL

- CYPXIB2
- cytochrome P-450Aldo
- cytochrome P-450C18
- cytochrome P450 11B2, mitochondrial
- cytochrome P450 11B2, mitochondrial precursor
- cytochrome P450, family 11, subfamily B, polypeptide 2
- cytochrome P450, subfamily XIB (steroid 11-beta-hydroxylase), polypeptide 2
- mitochondrial cytochrome P450, family 11, subfamily B, polypeptide 2
- P-450C18
- P450aldo
- P450C18
- steroid 11-beta-monooxygenase
- steroid 11-beta/18-hydroxylase
- steroid 18-hydroxylase, aldosterone synthase, P450C18, P450aldo

Additional Information & Resources

Educational Resources

- Endocrinology (2001): Synthesis of the Major Steroid Hormones Secreted by the Adrenal Cortex
<https://www.ncbi.nlm.nih.gov/books/NBK26/?rendertype=box&id=A463>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CYP11B2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- CYTOCHROME P450, SUBFAMILY XIB, POLYPEPTIDE 2
<http://omim.org/entry/124080>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CYP11B2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CYP11B2%5Bgene%5D>
- HGNC Gene Family: Cytochrome P450 family 11
<http://www.genenames.org/cgi-bin/genefamilies/set/1007>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2592
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1585>
- UniProt
<http://www.uniprot.org/uniprot/P19099>

Sources for This Summary

- Amir O, Amir RE, Paz H, Mor R, Sagiv M, Lewis BS. Aldosterone synthase gene polymorphism as a determinant of atrial fibrillation in patients with heart failure. *Am J Cardiol.* 2008 Aug 1;102(3):326-9. doi: 10.1016/j.amjcard.2008.03.063. Epub 2008 May 29.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18638595>
- OMIM: CYTOCHROME P450, SUBFAMILY XIB, POLYPEPTIDE 2
<http://omim.org/entry/124080>
- Connell JM, Fraser R, MacKenzie SM, Friel EC, Ingram MC, Holloway CD, Davies E. The impact of polymorphisms in the gene encoding aldosterone synthase (CYP11B2) on steroid synthesis and blood pressure regulation. *Mol Cell Endocrinol.* 2004 Mar 31;217(1-2):243-7. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15134824>
- Martinez-Aguayo A, Fardella C. Genetics of hypertensive syndrome. *Horm Res.* 2009;71(5):253-9. doi: 10.1159/000208798. Epub 2009 Apr 1. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19339789>
- Moraitis AG, Rainey WE, Auchus RJ. Gene mutations that promote adrenal aldosterone production, sodium retention, and hypertension. *Appl Clin Genet.* 2013 Dec 24;7:1-13. doi: 10.2147/TACG.S35571. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24399884>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3882136/>
- Nguyen HH, Hannemann F, Hartmann MF, Malunowicz EM, Wudy SA, Bernhardt R. Five novel mutations in CYP11B2 gene detected in patients with aldosterone synthase deficiency type I: Functional characterization and structural analyses. *Mol Genet Metab.* 2010 Aug;100(4):357-64. doi: 10.1016/j.ymgme.2010.04.016. Epub 2010 May 21.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20494601>
- Quack I, Vonend O, Rump LC. Familial hyperaldosteronism I-III. *Horm Metab Res.* 2010 Jun;42(6):424-8. doi: 10.1055/s-0029-1246187. Epub 2010 Feb 3. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20131203>

- Stowasser M, Gordon RD. Familial hyperaldosteronism. J Steroid Biochem Mol Biol. 2001 Sep; 78(3):215-29. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11595502>
- White PC. Aldosterone synthase deficiency and related disorders. Mol Cell Endocrinol. 2004 Mar 31;217(1-2):81-7. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15134805>
- Williams SS. Advances in genetic hypertension. Curr Opin Pediatr. 2007 Apr;19(2):192-8. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17496764>
- Yu Y. The CYP11B2 -344C/T variant is associated with ischemic stroke risk: An updated meta-analysis. J Renin Angiotensin Aldosterone Syst. 2015 Jun;16(2):382-8. doi: 10.1177/1470320313492362. Epub 2013 Jun 7.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23748625>

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/CYP11B2>

Reviewed: April 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services